# infantile systemic hyalinosis

Infantile systemic hyalinosis is a disorder that severely affects many areas of the body, including the skin, joints, bones, and internal organs. Hyalinosis refers to the abnormal accumulation of a clear (hyaline) substance in body tissues. The signs and symptoms of this condition are present at birth or develop within the first few months of life. Infantile systemic hyalinosis is characterized by painful skin bumps that frequently appear on the hands, neck, scalp, ears, and nose. They also develop in joint creases and the genital region. These skin bumps may be large or small and often increase in number over time.

Lumps of noncancerous tissue also form in the muscles and internal organs of children with infantile systemic hyalinosis, causing pain and severe complications. Most affected individuals develop a condition called protein-losing enteropathy due to the formation of lumps in their intestines. This condition results in severe diarrhea, failure to gain weight and grow at the expected rate (failure to thrive), and general wasting and weight loss (cachexia).

Infantile systemic hyalinosis is also characterized by overgrowth of the gums (gingival hypertrophy). Additionally, people with this condition have joint deformities (contractures) that impair movement. Affected individuals may grow slowly and have bone abnormalities.

Although children with infantile systemic hyalinosis have severe physical limitations, mental development is typically normal. Affected individuals often do not survive beyond early childhood due to chronic diarrhea and recurrent infections.

## Frequency

The prevalence of infantile systemic hyalinosis is unknown. Fewer than 20 people with this disorder have been reported.

## Genetic Changes

Mutations in the *ANTXR2* gene (also known as the *CMG2* gene) cause infantile systemic hyalinosis. The *ANTXR2* gene provides instructions for making a protein involved in the formation of tiny blood vessels (capillaries). Researchers believe that the ANTXR2 protein is also important for maintaining the structure of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues.

The signs and symptoms of infantile systemic hyalinosis are caused by the accumulation of a hyaline substance in different parts of the body. The nature of this

substance is not well known, but it is likely made up of protein and sugar molecules. Researchers suspect that mutations in the *ANTXR2* gene disrupt the formation of basement membranes, allowing the hyaline substance to leak through and build up in various body tissues.

## Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Other Names for This Condition

- inherited systemic hyalinosis

## Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hyaline fibromatosis syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2745948/

Other Diagnosis and Management Resources

- GeneReview: Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525
- MedlinePlus Encyclopedia: Protein-losing enteropathy
  https://medlineplus.gov/ency/article/007338.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

## Additional Information & Resources

MedlinePlus

- Encyclopedia: Protein-losing enteropathy
  https://medlineplus.gov/ency/article/007338.htm
- Health Topic: Gum Disease
  https://medlineplus.gov/gumdisease.html
- Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html

Genetic and Rare Diseases Information Center

- Hyaline fibromatosis syndrome
  https://rarediseases.info.nih.gov/diseases/6807/hyaline-fibromatosis-syndrome

Educational Resources

- Orphanet: Infantile systemic hyalinosis
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2176

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/derm.html

GeneReviews

- Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28infantile+systemic+hyalinosis%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- HYALINE FIBROMATOSIS SYNDROME
  http://omim.org/entry/228600

## Sources for This Summary


- Antaya RJ, Cajaiba MM, Madri J, Lopez MA, Ramirez MC, Martignetti JA, Reyes-Múgica M. Juvenile hyaline fibromatosis and infantile systemic hyalinosis overlap associated with a novel mutation in capillary morphogenesis protein-2 gene. Am J Dermatopathol. 2007 Feb;29(1):99-103.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/17284973
- Dowling O, Difeo A, Ramirez MC, Tukel T, Narla G, Bonafe L, Kayserili H, Yuksel-Apak M, Paller AS, Norton K, Teebi AS, Grum-Tokars V, Martin GS, Davis GE, Glucksman MJ, Martignetti JA. Mutations in capillary morphogenesis gene-2 result in the allelic disorders juvenile hyaline fibromatosis and infantile systemic hyalinosis. Am J Hum Genet. 2003 Oct;73(4):957-66. Epub 2003 Sep 12.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/12973667
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180616/
- GeneReview: Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525
- Hanks S, Adams S, Douglas J, Arbour L, Atherton DJ, Balci S, Bode H, Campbell ME, Feingold M, Keser G, Kleijer W, Mancini G, McGrath JA, Muntoni F, Nanda A, Teare MD, Warman M, Pope FM, Superti-Furga A, Futreal PA, Rahman N. Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. Am J Hum Genet. 2003 Oct;73(4):791-800. Epub 2003 Aug 21.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/14508707
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180602/
- Lindvall LE, Kormeili T, Chen E, Ramirez MC, Grum-Tokars V, Glucksman MJ, Martignetti JA, Zaragoza MV, Dyson SW. Infantile systemic hyalinosis: Case report and review of the literature. J Am Acad Dermatol. 2008 Feb;58(2):303-7. doi: 10.1016/j.jaad.2007.06.008. Review.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/18222328


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